



RGS9BP gene

regulator of G-protein signaling 9 binding protein

Normal Function

The *RGS9BP* gene (which is also known as *R9AP*) provides instructions for making a protein called RGS9 binding protein. This protein is found in the retina, which is the specialized tissue at the back of the eye that detects light and color. Within the retina, the protein is associated with light-detecting cells called photoreceptors. As its name suggests, RGS9 binding protein interacts with a protein called RGS9 (which is produced from the *RGS9* gene). It anchors the RGS9 protein to photoreceptors and stimulates RGS9's activity.

When light enters the eye, it stimulates specialized pigments in photoreceptor cells. This stimulation triggers a series of chemical reactions that produce an electrical signal, which is interpreted by the brain as vision. (This process is known as phototransduction.) Once photoreceptors have been stimulated by light, they must return to their resting state before they can be stimulated again. RGS9 and RGS9 binding protein are involved in a chemical reaction that helps return photoreceptors to their resting state quickly after light exposure.

Health Conditions Related to Genetic Changes

bradyopsia

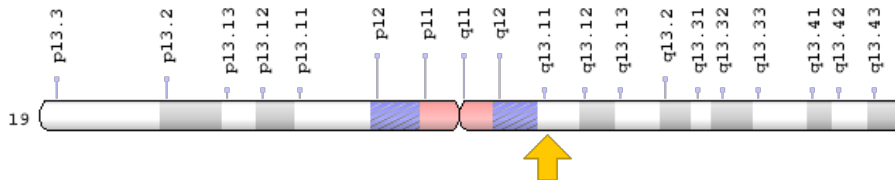
At least six mutations in the *RGS9BP* gene have been found to cause bradyopsia, a rare condition that affects vision. In people with bradyopsia, the eyes adapt more slowly than usual to changing light conditions (for example, walking out of a darkened movie theater into daylight or driving into a dark tunnel on a sunny day). Some affected individuals also have difficulty seeing small moving objects, such as a tennis ball.

The *RGS9BP* gene mutations that cause bradyopsia prevent RGS9 binding protein from anchoring the RGS9 protein to photoreceptor cells. The resulting loss of RGS9 protein function prevents photoreceptors from recovering quickly after responding to light. Normally they return to their resting state in a fraction of a second, but in people with *RGS9BP* gene mutations, it can take ten seconds or longer. During that time, the photoreceptors cannot respond to light. This delay causes temporary blindness in response to changing light conditions and may interfere with seeing small objects when they are in motion.

Chromosomal Location

Cytogenetic Location: 19q13.11, which is the long (q) arm of chromosome 19 at position 13.11

Molecular Location: base pairs 32,675,407 to 32,678,300 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FLJ45744
- R9AP
- regulator of G-protein signaling 9-binding protein
- regulator of G protein signaling 9 binding protein
- RGS9 anchor protein
- RGS9-anchoring protein

Additional Information & Resources

Educational Resources

- Webvision: The Organization of the Retina and Visual System (2010): Phototransduction in Rods and Cones
<https://www.ncbi.nlm.nih.gov/books/NBK52768/>
- Webvision: The Organization of the Retina and Visual System (updated 2012): Photoreceptors
<https://www.ncbi.nlm.nih.gov/books/NBK11522/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28RGS9BP%5BTIAB%5D%29+OR+%28R9AP%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- REGULATOR OF G PROTEIN SIGNALING 9-BINDING PROTEIN
<http://omim.org/entry/607814>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RGS9BP%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=30304
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/388531>
- UniProt
<http://www.uniprot.org/uniprot/Q6ZS82>

Sources for This Summary

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Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17698770>
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- Nishiguchi KM, Sandberg MA, Kooijman AC, Martemyanov KA, Pott JW, Hagstrom SA, Arshavsky VY, Berson EL, Dryja TP. Defects in RGS9 or its anchor protein R9AP in patients with slow photoreceptor deactivation. Nature. 2004 Jan 1;427(6969):75-8.
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